In-Depth

Overview

Points To Remember About Heritable Disorders of Connective Tissue

- There are more than 200 heritable disorders that can affect the tissues between the cells of your body that give tissues form and strength.
- All of these diseases are related to problems in genes that are responsible for building connective tissues.
- Some heritable disorders of connective tissue change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work.
- Treatments can include regular check-ups with your doctor as well as medicines or nutritional supplements.
- A nutritious diet, exercise, and healthy lifestyle habits can also help.

Heritable disorders of connective tissue are a family of more than 200 conditions that affect tissues between cells that give tissues form and strength. All of these diseases are directly related to problems in genes that are responsible for building connective tissues.

Some other connective tissue problems are not directly linked to changes in tissue-building genes. Many, but not all, of the disorders are rare.

What happens in heritable disorders of connective tissue?

Some gene defects alter the proteins that make up connective tissue. This can alter normal function of connective tissue, which usually supports many parts of the body by:

- Bringing nutrients to the tissue.
- Giving tissue form and strength.
- Helping some of the tissues do their work.

Who Gets

If you have a heritable disorder of connective tissue, you inherited an altered gene either from one or from both parents. This defective gene affects functioning of connective tissues.

Heritable disorders of connective tissue can affect people of all racial and ethnic groups. All ages and both sexes are affected. Many of these disorders are rare. Some may not be seen at birth, but only appear after a certain age or after exposure to a particular environmental stress.

You are more likely to have the disease if someone in your family has had the disease, or if one of your parents have the gene for the disorder.

Types

Some common heritable disorders of connective tissue include:

- **Ehlers-Danlos syndrome** can be caused by defects in several genes. In most cases, the gene defect involves collagen, the major protein-building material of bone. People with this syndrome have loose joints; fragile, small blood vessels; abnormal scar formation and wound healing; and soft skin that stretches excessively but returns to normal after being pulled. Some forms can cause problems with the eyes, spine, and internal organs.
- **Epidermolysis bullosa** affects the skin, causing it to blister easily. Some people may have just a few blisters on skin, whereas others may have many blisters. In some forms, blisters may form in the mouth, stomach, esophagus, bladder, and other parts of the body.
- **Marfan syndrome** is caused by a defect in the gene that produces a protein important to connective tissue. People with Marfan syndrome tend to have a tall, thin build with long arms and legs and “spider-like” fingers. Other problems include a sideways curve of the

spine; crowded teeth; flat feet; abnormal position of the eye's lens; and enlargement of the beginning part of the aorta, the major vessel carrying blood away from the heart.
- **Osteogenesis imperfecta** is caused by a defect in the gene that makes collagen. People with this disorder have fragile bones, low muscle mass, and loose joints and ligaments. Some people may have a blue or gray tint to the whites of the eyes, thin skin, growth deficiencies, and fragile teeth. A curved spine, breathing problems and hearing loss may also develop.

Symptoms

Symptoms of heritable disorders of connective tissue can include:

- **Bone growth problems:** Bones can be brittle, too long, or too short.
- **Joint issues:** Joints can be too loose or too tight.
- **Skin problems:** Skin may be loose, hang in folds, or easily blister.
- **Blood vessel damage:** Blood vessels may be weak or become blocked.
- **Height issues:**You may be unusually tall or short.
- **Head and facial structural problems:** The head and face may look different from others.

Diagnosis

Your doctor may do the following to diagnose you with heritable disorders of connective tissue:

- Medical history to determine system and ask about family history of the disease.
- Physical examination.
- Laboratory tests to confirm the disease.

You may wish to seek genetic counseling if you wish to have a child. A genetic counselor can help you estimate the risk of having a child with the disease. The genetic counselor can also tell you about certain tests that:

- Determine if you have certain altered genes that could be passed to your children.
- Screen your newborn for the disease.
- Test your fetus early in pregnancy to look for an altered form of a gene.

Treatment

Heritable disorders of connective tissue are a wide range of disorders, each requiring a specific program for management and treatment. Treatments can include:

- **Regular monitoring** to assess tissue changes.

- **Metabolic treatments** such as:
  - Vitamin B6 to correct a liver enzyme problem.
  - Drugs to slow the widening of the aorta.
  - Drugs to strengthen brittle bones.

Living With

Maintaining general health is important if you have a heritable disorder of connective tissue. You should talk to your doctor about a plan that includes:

- A nutritious diet.
- Exercise.
- Healthy lifestyle habits.

Research Progress

Scientists supported by the National Institutes of Health are working to better understand heritable disorders of connective tissue by:

- Examining how gene alterations produce the disorders.
- Developing methods to grow skin tissue for people affected by a life-threatening skin disease.
- Forming the Brittle Bone Disorders Consortium of the Rare Disease Clinical Research Network, a collaboration among researchers to better understand rare diseases characterized by fragile bones.
- Studying connective tissue defects in mice and humans.
- Studying aneurysms (weak spots in blood vessel walls that threaten to burst).

Related Resources

U.S. Food and Drug Administration
Toll free: 888-INFO-FDA (888-463-6332)
Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics
Website: https://www.cdc.gov/nchs

American Academy of Orthopaedic Surgeons

Website: https://www.aaos.org

American Academy of Dermatology
Website: https://www.aad.org

Coalition for Heritable Disorders of Connective Tissue
Website: https://www.chdct2.org

Genetic Alliance
Website: https://www.geneticalliance.org

National Organization for Rare Disorders
Website: https://www.rarediseases.org

National Society of Genetic Counselors
Website: https://www.nsgc.org

Dystrophic Epidermolysis Bullosa Research Association of America, Inc.
Website: https://www.debra.org

Ehlers-Danlos National Foundation
Website: https://www.ednf.org/

National Association for Pseudoxanthoma Elasticum
Website: https://www.pxenape.org

National Marfan Foundation
Website: https://www.marfan.org

Osteogenesis Imperfecta Foundation
Website: https://www.oif.org

PXE International
Website: https://www.pxe.org

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- Asian Language Health Information
- Spanish Language Health Information

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Related Information

Epidermólisis Ampollosa: Esenciales: hojas informativas de fácil lectura
Marfan: Esenciales: hojas informativas de fácil lectura
Osteogénesis Imperfecta: Esenciales: hojas informativas de fácil lectura
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